# GSN gene

gelsolin

## Normal Function

The *GSN* gene provides instructions for making two forms of a protein called gelsolin. One form remains inside the cell (cellular gelsolin) and the other form is released from the cell (secreted gelsolin). Both forms of the gelsolin protein attach (bind) to another protein called actin. Actin proteins are organized into filaments, which form a network (the cytoskeleton) that gives structure to cells and allows them to change shape and move. Gelsolin helps assemble or disassemble actin filaments. It is thought that, through this function, the gelsolin protein regulates the formation of the actin cytoskeleton.

## Health Conditions Related to Genetic Changes

lattice corneal dystrophy type II

At least two mutations in the *GSN* gene cause lattice corneal dystrophy type II. This condition is characterized by the accumulation of protein clumps called amyloid deposits in many tissues throughout the body, including the clear, outer covering of the eye (the cornea); the skin; and the nerves. These protein clumps contain the gelsolin protein.

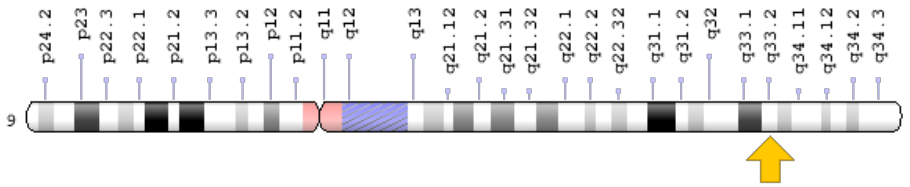
*GSN* gene mutations that cause lattice corneal dystrophy type II change a single protein building block (amino acid) in the gelsolin protein: the amino acid aspartic acid at position 187. The most common mutation replaces the aspartic acid with the amino acid asparagine (written as Asp187Asn or D187N). Another mutation replaces the aspartic acid with the amino acid tyrosine (written as Asp187Tyr or D187Y).

The amino acid change is found in both the cellular and secreted forms of the gelsolin protein. However, only the secreted form of the protein is involved in the amyloid deposits. The altered gelsolin protein is broken down differently than the normal protein, which results in an abnormal gelsolin protein fragment that is released from the cell. These protein fragments accumulate and form amyloid deposits. Amyloid deposits in the eyes, skin, and nerves lead to the signs and symptoms of lattice corneal dystrophy type II, such as vision impairment; paralysis of facial muscles; and thick, sagging skin.

## Chromosomal Location

Cytogenetic Location: 9q33.2, which is the long (q) arm of chromosome 9 at position 33.2

Molecular Location: base pairs 121,201,082 to 121,332,844 on chromosome 9 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)




Credit: Genome Decoration Page/NCBI

## Other Names for This Gene

- actin-depolymerizing factor
- ADF
- AGEL
- brevin
- DKFZp313L0718
- GELS_HUMAN
- gelsolin isoform a precursor
- gelsolin isoform b
- gelsolin isoform c

## Additional Information & Resources

Educational Resources

- Molecular Biology of the Cell (fourth edition, 2002): Severing Proteins Regulate the Length and Kinetic Behavior of Actin Filaments and Microtubules https://www.ncbi.nlm.nih.gov/books/NBK26809/#A3028

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28GSN%5BTIAB%5D%29+OR+%28gelsolin%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1080+days%22%5Bdp%5D

OMIM

- GELSOLIN
  http://omim.org/entry/137350

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_GSN.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=GSN%5Bgene%5D
- HGNC Gene Family: Gelsolin/villins
  http://www.genenames.org/cgi-bin/genefamilies/set/950
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=4620
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/2934
- UniProt
  http://www.uniprot.org/uniprot/P06396

## Sources for This Summary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